Phenylketonuria/ Hyperphenylalaninemia

What is newborn screening?

Newborn screening is a blood test to check for conditions that might be hidden at birth. To do the screening, a nurse takes a few drops of blood from your baby's heel soon after birth. This blood sample is required for all newborn babies.

Newborn screening is not the same as diagnostic testing. A diagnostic test can tell with more certainty whether or not a child has a condition. On the other hand, a screening test simply indicates that a child ***may*** have a condition. The purpose of a screening test is to find babies that should have diagnostic testing. When a child with an out-of-range newborn screening result has a follow-up test result within the normal range, it is sometimes called a "false positive."

KEY POINTS:

- **You have just heard that your baby may have PKU or H-PHE. Please understand that the newborn screening is just that---a screening test. Further testing is required to confirm or rule out the diagnosis.**
- **Many babies who have out of range newborn screens are healthy, and will not be diagnosed with PKU or H-PHE.**
- **If treated early, children with PKU and H-PHE can have healthy growth and development.**

What if my baby needs more testing?

If you are told that your baby needs follow-up testing, it does not necessarily mean that your baby is at risk. An out of range result may occur because:

- The sample was too small
- The sample was collected too early
- The sample was collected too close to a feeding
- The baby was born too early or had a low birth weight

Many babies who have follow up testing for phenylketonuria and hyperphenylalaninemia are healthy, and will not be diagnosed with PKU or H-PHE. However, out of range screening results CAN indicate a disorder, so it is important to follow your doctor's advice & get your baby tested quickly so that final results can be confirmed.

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What is phenylketonuria and hyperphenylalaninemia?

Phenylketonuria (PKU) is a condition in which the body cannot break down one of the amino acids found in proteins. PKU is considered an amino acid condition because people with PKU cannot break down the amino acid called phenylalanine. If left untreated, PKU can be a very serious condition. However, if the condition is detected early and treatment is begun, individuals with PKU can lead healthy lives.

PKU is a condition with multiple forms, each of which have different treatments and outcomes. Benign hyperphenylalaninemia (H-PHE) is a mild form of phenylketonuria. Most people with this condition experience mild symptoms or no symptoms.

What does this mean?

Although PKU cannot be cured, it can be treated. If further testing finds that your baby has PKU, he or she will have to stay on a low protein diet throughout life. This will help to prevent health issues. It is recommended that treatment be started as soon as possible.

What happens next?

Your baby's doctor may ask for your baby to have more testing. This follow up testing is important to know if treatment is needed. In some cases, you may be asked to visit a healthcare specialist. The specialist may want to switch your baby to a special formula since breastmilk and milk-based formulas contain protein.

What are the signs and symptoms of PKU and H-PHE?

Signs and symptoms of PKU can be very different from one baby to another. Some babies do not show any symptoms. Other babies have dry, scaly or pale skin, "musty" body odor, or irritability. Most babies with H-PHE don't show any signs. If you become concerned about your baby's growth, feeding or activity, please talk to your pediatrician.

What if I still have questions?

We understand that this can be an overwhelming and emotional process. Many families have questions and concerns. The Connecticut Newborn Diagnosis and Treatment Network (the Network) is available to put you in touch with the best resource. To reach the Network, you can call 860-837-7870, Monday-Friday, 8:30am-4:30pm. We also recommend the website www.babysfirsttest.org as an accurate and informative resource.

This fact sheet was written for information purposes only. It should not replace medical advice, diagnosis or treatment.

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